TABLE 1 Exon-Intron Boundary Sequences of the Human alpha-7 nAChR Subunit Gene

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| Exon Number | Exon length(bp) | cDNA position | splice acceptor | flanking exon sequence | splice donor | Intron number | Intron Size approximate (Kb) |
|----------------|--------------------|------------------|--------------------|------------------------|-----------------|------------------|---------------------------------|
| 1 | 55 | 1-55 | | CTG CAC G L H | GTAAAGCCAC | 1 | 0.3 |
| 2 | 140 | 56-195 | TCTCCTTAAG | TG TCCGAC GTG | GTGAGTCCCG | 2 | Unknown |
| 3 | 45 | 196-240 | TTTTTTGAAG | GAT GAGCAA ATG D E O M | GTAAGTTAAG | 3 | 9.0 |
| 4 | 110 | 241-350 | TGTGTGTCAG | TCT TGGAAC AG | GTAAGCATAT | 4 | Unknown |
| 5 | | 351-430 | CTGTTTCTAG | T GCT GATCCT CCA G | GTAAGCTGCA | 5 | 4.0 |
| 6 | 168 | 431-598 | ACCCACACAG | GC ATACTA GTG G | GTAAGCCATG | 6 | 1.0 |
| 7 | 195 | 599-793 | CCCTATGGAG | | GTAAGCGCCC | 7 | 1.0 |
| 8 | 87 | 794-880 | TATGTTTTAG | GG ATATTG ATA G | GTAAGGCAAG | 8 | 3.5 |
| 9 | 110 | 881-990 | CTCTCCACAG | CC CAGAAG TGG | GTACGTTCCT | 9 | 5.0 |
| 10 | 519 | 991-1509 | GTCTCCCCAG | A Q K W ACC AGA T R | | | |

FIGURE 2

TABLE 2 Sequence Variants Identified in Full-Length and Duplicated Genomic Clones

| CHRIS 5-10 HYBRID 1-10 YAC D-948a10 5-10 D-853b12 6-10 D/F 5-10 | | + TG | | | | |)) | | | 1269 | _ | 1335 | |
|---|-----|------|--------|-----|------|---|--------|-----|-----|------|-------------|------|------------------|
| | | | | Ö | C/T | | G/A | | 2/2 | | 2/2 | | 6GT |
| + | | | | | | | | | | | | | 500 |
| - | | | ن 1 | + | E- | | | - | | | , | | |
| - | | | -TG | - | · [= | | | (4 | ی د | | ی ار | | 199 199 |
| | _ | +TG | | C/T | Ţ, | | G/A | | + | C/T | ,)/) | | 100 |
| + | ┪ | -TG | | | | • | | | | |)) | | , E |
| F-134h10 1-10 | +TG | | | U | | ß | _ | | U | | C | - | F (2) |
| F-776a12 1-10 | +TG | | | U | | G | | | U | - | | | ָבָּטָ בַּטָּ |
| F-791e6 1-10 | +TG | | | U | | U | | - | U | | | | 100 100 |
| F-811b6 1-10 | +TG | | | U | | G | | + | U | + |) (| + | ٠ د د د |
| F-953g6 1-10 | +TG | | | U | | b | | - | U | |) (| - | 100 |
| F-859c11 1-10 | +TG | | | O | | υ | | | 0 | _ |) c | - | 500 |
| F-810f11 1-10 | +TG | | | U | | O | | + | U | |) (| | 1 E |
| F-801el 1-10 | +TG | | | U | | U | | + | U | - |) (| | 100 |
| | | | | - | + | | | 1 | | 1 | , , + | | 1500 |
| BAC | | | | - | - | | | | + | | | | |
| F-467018 1-10 | +TG | | | U | - | ပ | | + | ľ | - | - | E | 800 |

| 0 | T/T 0 | | | | |
|-----------------------|----------------------------|--|--|--|--|
| EXON 10 1335 | 1 24 19 | | | | |
| | C/C 24 | | | | |
| 0 | 1/1 | | | | |
| EXON 10 1269 | C/T 36 | | | | |
| | T G/G G/A A/A C/C 0 43 0 6 | | | | |
| 7 | A/A 0 | | | | |
| EXON 690 | G/A 43 | | | | |
| | 0/0 | | | | |
| 7 | /H | | | | |
| EXON 654 | C/C C/T 5 38 | | | | |
| | c/c 5 | | | | |
| 6 -498 | -/- | | | | |
| EXON 6 +/- 497-498 | 33 | | | | |
| | 10 | | | | |
| Control # | 43 | | | | |
| DNA | Control Genomic DNA | | | | |

TABLE 3
Expression Analysis of Sequence Variants

| 1-10 cDNA | 5-10 cDNA | DNA | 1-10 cDNA | 5-10 cDNA | DNA | 1-10 | 5-10 | | 1 40 | I | | | T | - | T | |
|--------------|---|---------------------------------------|--------------------|-----------------------------------|---|--|--|---|---|---|---|---|--------------------------------|--|---|---|
| | | | | 1 ~ | 1 | cDNA | cDNA | DNA | 1-10 cDNA | 5-10 cDNA | DNA | 1-10 cDNA | 5-10 cDNA | DNA | 1-10 cDNA | 5-10 cDNA |
| . • | +/- | CT | С | CT | GA | G | GA | G | G | G | СТ | CT | СТ | c | С | С |
| + | + | С | С | С | GA | | | | | | | - 6 | | _ | | - c |
| + | +/- | CT | С | CT | | | | | | | | - | | _ | 1 | |
| + | + | CT | c | | | | | | | | | - <u>CI</u> | | | | CT |
| + | + | С | | | | | | | | | | <u>\</u> | 1 | | <u> </u> | C |
| + | +/- | | | | | | | | | | <u> </u> | CI | | _ | | C |
| | + | + + + + + + + + + + + + + + + + + + + | + +/- CT + + CT | + +1- CT C + + CT C + + C C | + +/- CT C CT + + CT C CT + + C C C | + +1- CT C CT GA + + CT C CT GA + + C C C GA | + +/- CT C CT GA G + + CT C CT GA G + + CT C CT GA G + + C C C GA GA | + +/- CT C CT GA G GA. + + CT C CT GA G GA. + + C C C CT GA G GA. | + +/- CT C CT GA G GA G + + CT C CT GA G GA G + + CT C CT GA G GA G + + CC C CT GA G GA GA GA | + +1- CT C CT GA G GA G G + + CT C CT GA G GA G G + + CT C CT GA G GA G G + + CC C C GA GA GA GA GA | + +1- CT C CT GA G GA G G G G G G G G G G G G G G G | + +/- CT C CT GA G GA G G CT + + CT C CT GA G GA G G CT + + CT C CT GA G GA G G G CT + + C C C C GA GA GA GA GA GA CT | + + C C C GA G GA G G CT C + + | + + C C C GA G GA G G CT C CT + +/- CT C CT GA G GA G G G CT CT CT + + CT C CT GA G GA G G CT CT CT + + CT C CT GA G GA GA GA GA CT CT CT | + + C C C GA G GA G G G CT C CT C + +/- CT C CT GA G GA G G G CT CT CT CT + + CT C CT GA G GA G G G CT CT CT CT + + CT C C GA GA GA GA GA GA CT CT CT CT | + + C C C GA G GA G G G CT C CT C C + +/- CT C CT GA G GA G G G CT |

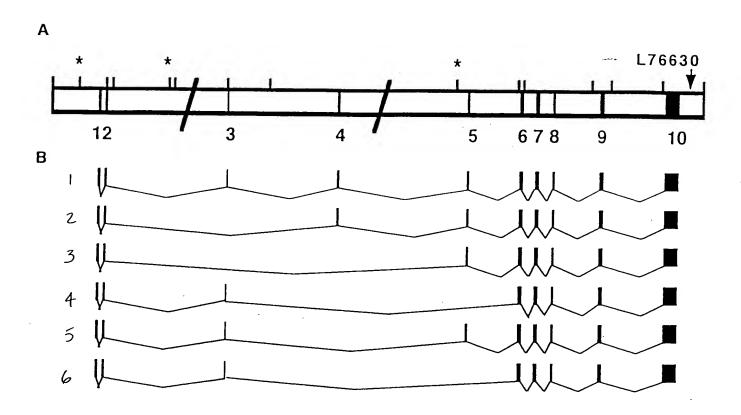


| 392 | agaacgcaag | ggagaggtag | agcctggcct | tgggcagccc | ctggcctggc | cagaggcgcg | aggccgagag |
|-----|------------|------------|------------|------------|------------|------------|------------|
| | | | | Al | P-2 | | |
| 322 | cccgctcggt | ggagactggg | ggtggaggtg | cccggagcgt | acccagcgcc | gggagtacct | cccgctcaca |
| 252 | | | | | | | ~~~~~~ |
| 232 | eccegggetg | cagtteeetg | ggtggccgcc | gagacgctgg | eeegggetgg | agggatggtg | gggcggggae |
| 182 | gggggcgggg | geggggeteg | tcacgtggag | aggcgcgcgg | gagaaagaa | ggegggggg | cgcgcccggc |
| | | | REB | 33 2 3 33 | Sp1 | | |
| 112 | tccttaaagg | cgcgcgagcc | gagcggcgag | gtgcctctgt | ggccgcaggc | gcaggcccgg | gcgacagccg |
| 40 | | | | _ | | | |
| -42 | agacgtggag | cgcgccggct | cgctgcagct | ccgggactca | | CTCGCCGGGA | GCCGTCTGGC |
| | | | | | Met | | |
| +29 | TGGCGCTGGC | CGCGTCGCTC | CTGCACGgta | aagccac | | | |

FIGURE 5

| | 1 | | | | | | |
|--------|--------------------|------------|------------|-------------|------------|------------|------------|
| EXON D | CAGGCCGCCA | CATAGCTCCC | GCCAAGTCCT | CGGTGCCCCT | TGCCATTTTC | CAGCCGCGTC | CCACGAGGGT |
| 297bp | CACGGCGGCG | GGGAGAGGTG | GAGCCGCGAG | AGCTCGGCCG | GGGGCCCCGC | CTGGTGGCCG | CGGCCATGAC |
| | AGCGGCTCGG | GACTGGCTCC | TTTTCCGCGC | CCCTCCCGCC | GGAGGTGAGG | GGAAGATGTC | CATGTCAGGG |
| | TTCAAGGCCA | AACCGAAGTT | ACTGGCCTCT | ATCTTCCAGG | AGAACCAGGA | GCCACAGCCG | CGGCTCACGC |
| | CCCACCGCAA | CATTAAGgtg | agtcgcc | • | | | |
| | | 297 | | | | | |
| | | 298 | | | | | |
| EXON C | ctc | atttcagATT | ACAAGTGGAC | ACCTGAGTCA | GCAGGACCTG | GAATCCCAGA | TGAGAGAGCT |
| 125bp | TATCTACACG | ACTCAGATCT | TGTTGTCACC | CCCATTATTG | ACAATCCAAA | GGTGCAGAAA | GCACTCTGAC |
| | AA gtgagttg | ta | | | | | |
| | 422 | | | | | | |
| | | 423 | | | | | |
| exon b | ttaaccac | agATAATGAA | ACAACCACCA | TCGGTTAAAT | TTGATGCAAA | AATATTGCAT | CTACCAGCAT |
| 64bp | TTTCAGgtag | gatcat | | | | | |
| | · 486 | | | | | | |
| | | 487 | | | | | |
| exon a | | ttctagTTCC | AATTGCTAAT | CCAGCATTTG | TGGATAGCTG | CAAACTGCGA | TATgtaagta |
| 47bp | aca | | | | | | 533 |
| | | | | | | | |
| | | 534 | | | | | |
| EXON 5 | ctgtttc | tagTGCTGAT | GAGCGCTTTG | ACGCCACATT | CCACACTAAC | GTGTTGGTGA | ATTCTTCTGG |
| 80bp | | GCATTGCCAG | TACCTGCCTC | CAGgtaagetg | ca | | |
| | | C 1 4 | | 613 | | | |
| | | 614 | 011010mmoo | | | | |
| EXON 6 | acccaca | cagGCATATT | CAAGAGTTCC | TGCTACATCG | | | |
| 27bp | | | | 640 | | | |

FIGURE 7



- 61 aggccgagag cccgctcggt ggagactggg ggtggaggtg cccggagcgt acccagcgcc
- 121 gggagtacct cccgctcaca cctcgggctg cagttccctg ggtggccgcc gagacgctgg
- 241 aggegegeg gggegggeg ggegggggeg egegeegge teettaaagg egegegagee
- 301 gagcggcgag gtgcctctgt ggccgcaggc gcaggcccgg gcgacagccg agacgtggag
- 361 egegeegget egetgeaget eegggaetea ac

- 1 agccctttcc caggcggtag cgggggcagt ggtgctgttg cccttttaaa ctgcggcttg
- 61 acgggagccg cgcctcctgt cggtggagtc ggttataaag ggagcagccc cgcaggccgc
- 121 cacatagete cegecaagte eteggtgeee ettgecattt teeageegeg eteceaegag
- 181 ggtcacggcg gcggggagag gtggagccgc gagagctcgg ccgggggccc cgcctggtgg
- 241 ccgcggccat gacagcggct cgggactggc tccttttccg cgcccctccc gccggaggtg
- 301 aggggaagat gtccatgtca gggttcaagg ccaaaccgaa gttactggcc tctatcttcc
- 361 aggagaacca ggagccacag ccgcggctca cgccccaccg caacattaag attacaagtg
- 421 gacacetgag teageaggae etggaateee agatgagaga gettatetae aegaeteaga
- 481 tettgttgte acceccatta ttgacaatee aaaggtgeag aaageaetet gacaatteea
- 541 attgctaatc cagcatttgt ggatagctgc aaactgcgat attgctgatg agcgctttga
- 601 egecacatte cacactaaeg tgttggtgaa ttettetggg cattgecagt acetgeetee
- 661 aggeatatte aagagtteet getacateg



1 caggeegeca catageteec gecaagteet eggtgeeect tgecatttte cageegeget

- 61 cccacgaggg tcacggcggc ggggagaggt ggagccgcga gagctcggcc gggggccccg
- 121 cetggtggce geggccatga cageggeteg ggaetggete etttteegeg ecceteeege
- 181 cggaggtgag gggaagatgt ccatgtcagg gttcaaggcc aaaccgaagt tactggcctc
- 241 tatetteeag gagaaceagg ageeacagee geggeteaeg eeceaeegea acattaagat
- 301 tacaagtgga cacctgagtc agcaggacct ggaatcccag atgagagagc ttatctacac
- 361 gactcagatc ttgttgtcac ccccattatt gacaatccaa aggtgcagaa agcactctga
- 421 caaataatga aacaaccacc atcggttaaa tttgatgcaa aaatattgca tctaccagca
- 481 ttttcagttc caattgctaa tccagcattt gtggatagct gcaaactgcg atattgctga
- 541 tgagcgcttt gacgccacat tccacactaa cgtgttggtg aattettetg ggcattgcca
- 601 gtacetgeet eeaggeatat teaagagtte etgetacate g